TECHNOLOGICAL INSTITUTE OF THE PHILIPPINES

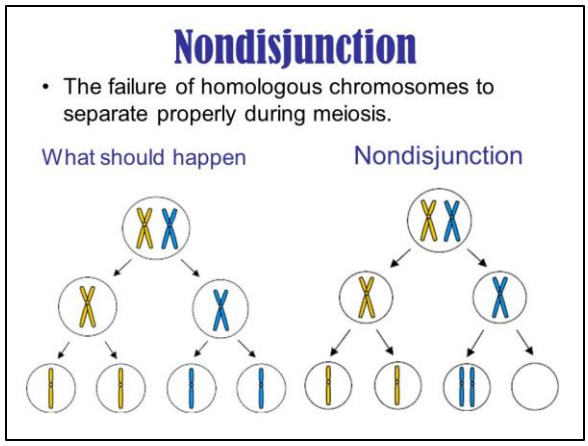
SCIENCE, TECHNOLOGY, ENGINEERING AND MATHEMATICS

HS BIO 001 – GENERAL BIOLOGY I

DISORDERS IN MITOSIS AND MEIOSIS

NON-DISJUNCTION

- is the failure of homologous chromosomes or sister chromatids to separate properly during cell division.
- It may appear during Mitosis, Meiosis I or in Meiosis II



ANEUPLOIDY *is a form of non disjunction*

- is the presence of an abnormal number of chromosomes in a cell, for example a human cell having 45 or 47 chromosomes instead of the usual 46.

HEMOPHILIA/ HAEMOPHILIA

- is a mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding.
- There are two main types of Haemophilia:

- Haemophilia A - which occurs because of not enough clotting factor VIII.

FACTOR VIII - is an essential blood-clotting protein, also known as anti-hemophilic factor (AHF). Factor VIII is produced in liver sinusoidal cells and endothelial cells outside of the liver throughout the body. This protein circulates in the bloodstream in an inactive form, bound to another molecule called von Willebrand factor, until an injury that damages blood vessels occurs.[6] In response to injury, coagulation factor VIII is activated and separates from von Willebrand factor. The active protein (sometimes written as coagulation factor VIIIa) interacts with another coagulation factor called factor IX. This interaction sets off a chain of additional chemical reactions that form a blood clot.

FVIII concentrated from donated blood plasma, or alternatively recombinant FVIIa can be given to hemophiliacs to restore **hemostasis** (**hemostasis** is the process which causes bleeding to stop, meaning to keep blood within a damaged blood vessel (the opposite of hemostasis is hemorrhage). It is the first stage of wound healing. This involves coagulation, blood changing from a liquid to a gel. Intact blood vessels are

central to moderating blood's tendency to form clots. Hemostasis has three major steps: 1) **vasoconstriction - the constriction of blood vessels, which increases blood pressure**;, 2) temporary blockage of a break by a platelet plug, and 3) blood coagulation, or formation of a fibrin clot. These processes seal the hole until tissues are repaired).

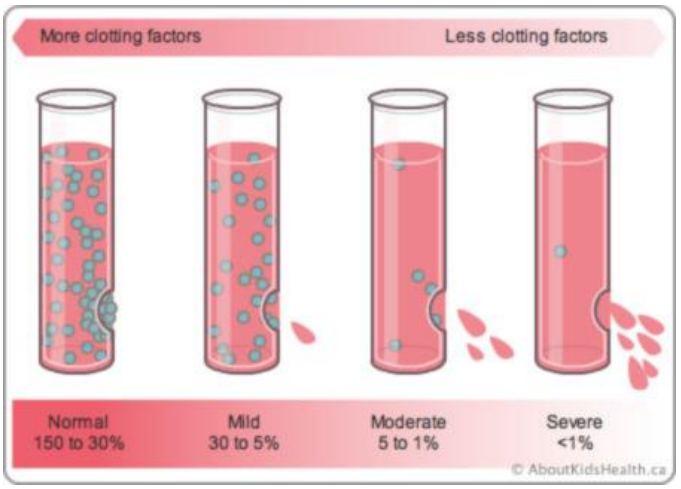
- Haemophilia B, which occurs due to not enough clotting factor IX.
  FACTOR IX - (or Christmas factor) is one of the serine proteases (are enzymes that cleave peptide bonds in proteins, in which serine serves as the Nucleophilic amino acid at the (enzyme's) active site.) of the coagulation system.

- Hemophilia is also called as "ROYAL DISEASE" because the royal family from England is a carrier of the disease
- Since Hemophilia is a sex linked trait, it resides in the X chromosomes. Male are victims and Females are carriers.
- Symptoms –
- Signs and symptoms of hemophilia vary, depending on your level of clotting factors. If your clotting-factor level is mildly reduced, you may bleed only after surgery or trauma. If your deficiency is severe, you may experience spontaneous bleeding.
- Signs and symptoms of spontaneous bleeding include:

  - Unexplained and excessive bleeding from cuts or injuries, or after surgery or dental work
  - Many large or deep bruises
  - Unusual bleeding after vaccinations
  - Pain, swelling or tightness in your joints
  - Blood in your urine or stool
  - Nosebleeds without a known cause
  - In infants, unexplained irritability

Emergency signs and symptoms of hemophilia include:

- Sudden pain, swelling and warmth in large joints, such as knees, elbows, hips and shoulders, and in your arm and leg muscles
- Bleeding from an injury, especially if you have a severe form of hemophilia
- Painful, prolonged headache
- Repeated vomiting
- Extreme fatigue
- Neck pain
- Double vision



People with Hemophilia: Prince Leopold (Left) and Queen Victoria (Right)

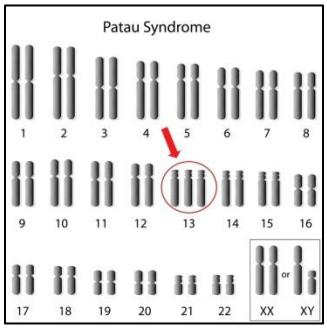
GENETIC SYNDROMES

A syndrome is a disease or disorder that has more than one identifying feature or symptom. Each particular genetic syndrome will have many typical features, depending on which aspects of development are affected by the abnormal genes or chromosomes.

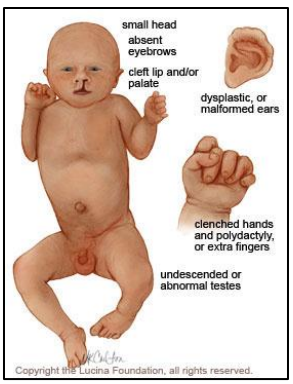
A child might be born with obvious body deformities, abnormal organ function (for example: heart, brain, gut, or kidney), or neurological problems (for example, when a baby's body is floppy or the baby is unable to nurse or bottle feed). However, many of the genetic syndromes start to take effect only once the baby has been born and is starting to feed and grow. These babies may look and act entirely normal at birth, but then develop problems later on in life.

SOME OF THE COMMON SYNDROMES ARE:

TRISOMY 13 or **PATAU'S SYNDROME**

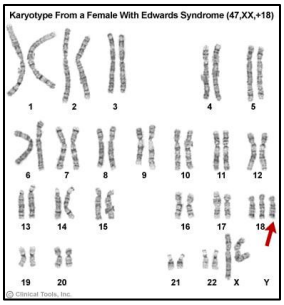


- is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Individuals withtrisomy 13 often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate), and weak muscle tone (hypotonia). Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within their first days or weeks of life. Only five percent to 10 percent of children with this condition live past their first year.

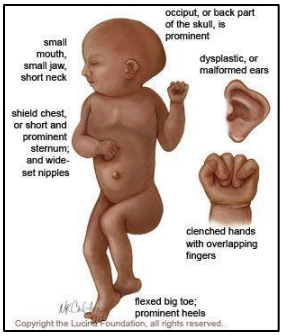


- Trisomy 13 occurs in about *1 in 16,000* newborns. Although women of any age can have a child with trisomy 13, the chance of having a child with this condition increases as a woman gets older.

TRISOMY 18 or **EDWARD'S SYNDROME**

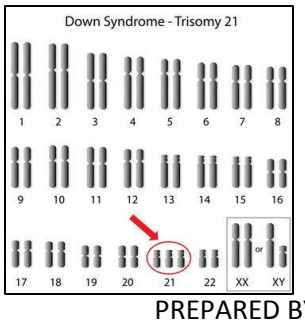


- is a chromosomal condition associated with abnormalities in many parts of the body. Individuals with trisomy 18 often have slow growth before birth (intrauterine growth retardation) and a low birth weight. Affected individuals may have heart defects and abnormalities of other organs that develop before birth. Other features of trisomy 18 include a small, abnormally shaped head; a small jaw and mouth; and clenched fists with overlapping fingers. Due to the presence of several life-threatening medical problems, many individuals with trisomy 18 die before birth or within their first month. Five to 10 percent of children with this condition live past their first year, and these children often have severe intellectual disability.



- Trisomy 18 occurs in about 1 in 5,000 live-born infants; it is more common in pregnancy, but many affected fetuses do not survive to term. Although women of all ages can have a child with trisomy 18, the chance of having a child with this condition increases as a woman gets older.

TRISOMY 21 or **DOWN'S SYNDROME**

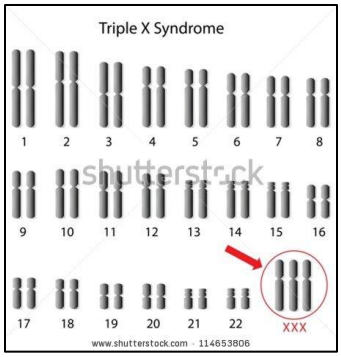


- a chromosomal condition that is associated with intellectual disability, a characteristic facial appearance, and weak muscle tone (hypotonia) in infancy. All affected

individuals experience cognitive delays, but the intellectual disability is usually mild to moderate.

- Down syndrome occurs in about 1 in 800 newborns. About 5,300 babies with Down syndrome are born in the United States each year, and approximately 200,000 people in this country have the condition. Although women of any age can have a child with Down syndrome, the chance of having a child with this condition increases as a woman gets older.

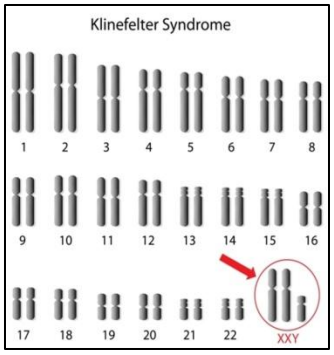
XXX SYNDROME OR TRIPLE X SYNDROME



- Triple X syndrome, also called trisomy X or 47,XXX, is characterized by the presence of an additional X chromosome in each of a female's cells. Although females with this condition may be taller than average, this chromosomal change typically causes no unusual physical features. Most females with triple X syndrome have normal sexual development and are able to conceive children.
- Triple X syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Delayed development of motor skills (such as sitting and walking), weak muscle tone (hypotonia), and behavioral and emotional difficulties are also possible, but these characteristics vary widely among affected girls and women. Seizures or kidney abnormalities occur in about 10 percent of affected females.

- This condition occurs in about 1 in 1,000 newborn girls. Five to 10 girls with triple X syndrome are born in the United States each day.

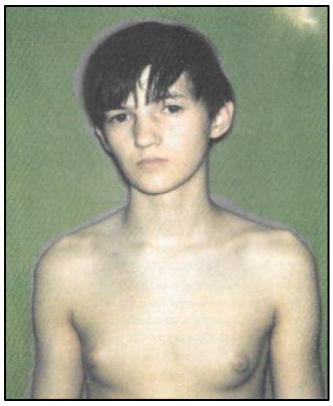
XXY SYNDROME OR KLINEFELTER'S SYNDROME



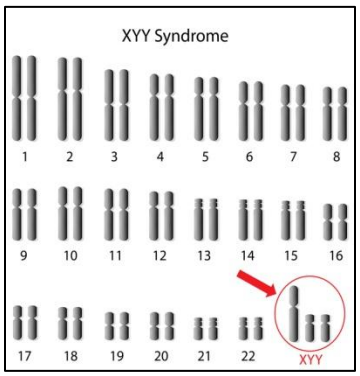
- Is a chromosomal condition that affects male physical and cognitive development. Its signs and symptoms vary among affected individuals.
- Affected individuals typically have small testes that do not produce as much testosterone as usual. Testosterone is the hormone that directs male sexual development before birth and during puberty. A shortage of testosterone can lead to delayed or incomplete puberty, breast enlargement (gynecomastia), reduced facial and body hair, and an inability to have biological children (infertility). Some affected individuals also have genital differences including undescended testes (cryptorchidism), the opening of the urethra on the underside of the penis (hypospadias), or an unusually small penis (micropenis).
- Older children and adults with Klinefelter syndrome tend to be taller than their peers. Compared with unaffected men, adults

with Klinefelter syndrome have an increased risk of developing breast cancer and a chronic inflammatory disease called systemic lupus erythematosus. Their chance of developing these disorders is similar to that of women in the general population.

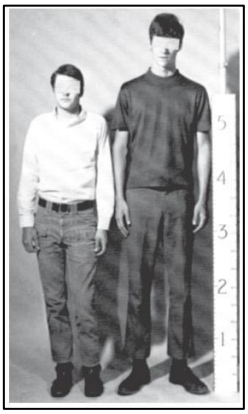
- Children with Klinefelter syndrome may have learning disabilities and delayed speech and language development. They tend to be quiet, sensitive, and unassertive, but personality characteristics vary among affected individuals.
- Klinefelter syndrome affects 1 in 500 to 1,000 newborn males. Most variants of Klinefelter syndrome are much rarer, occurring in 1 in 50,000 or fewer newborns.
- Researchers suspect that Klinefelter syndrome is underdiagnosed because the condition may not be identified in people with mild signs and symptoms. Additionally, the features of the condition v ary and overlap significantly with those of other conditions.



XYY SYNDROME OR JACOB'S SYNDROME

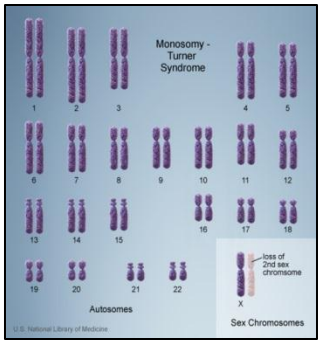


- characterized by an extra co py of the Y chromosome in each of a male's cells. Although males with this condition may be taller than average, this chromosomal change typically causes no unusual physical features. Most males with 47,XYY syndrome have normal sexual development and are able to father children.
- 47,XYY syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Delayed development of motor skills (such as sitting and walking), weak muscle tone (hypotonia), hand tremors or other involuntary movements (motor tics), and behavioral and emotional difficulties are also possible. These characteristics vary widely among affected boys and men.
- A small percentage of males with 47,XYY syndrome are diagnosed with autistic spectrum disorders, which are developmental conditions that affect communication and social interaction.



- This condition occurs in about 1 in 1,000 newborn boys. Five to 10 boys with 47,XYY syndrome are born in the United States each day.

XO SYNDROME OR TURNER'S SYNDROME

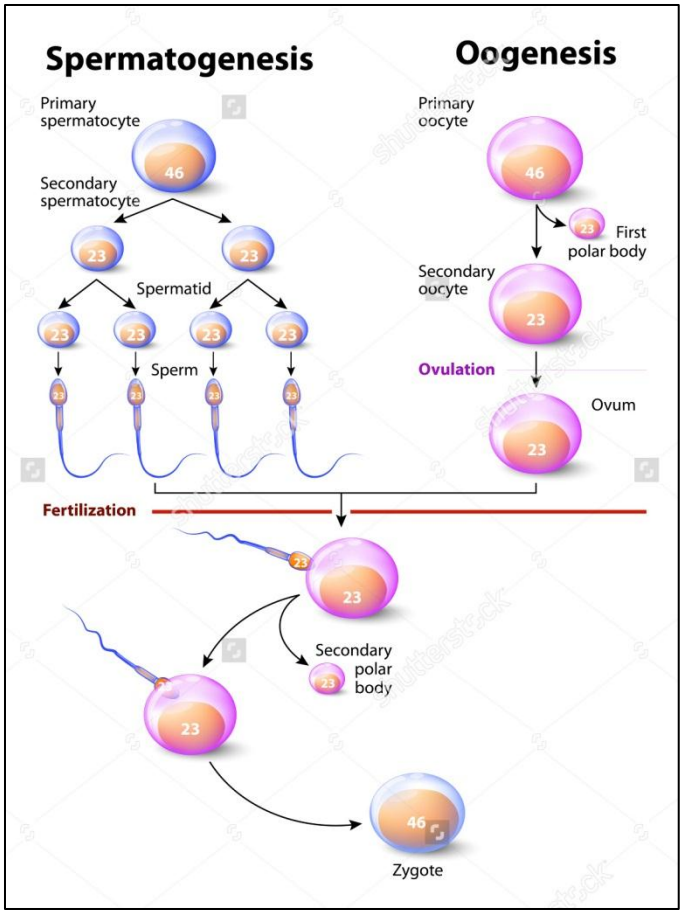


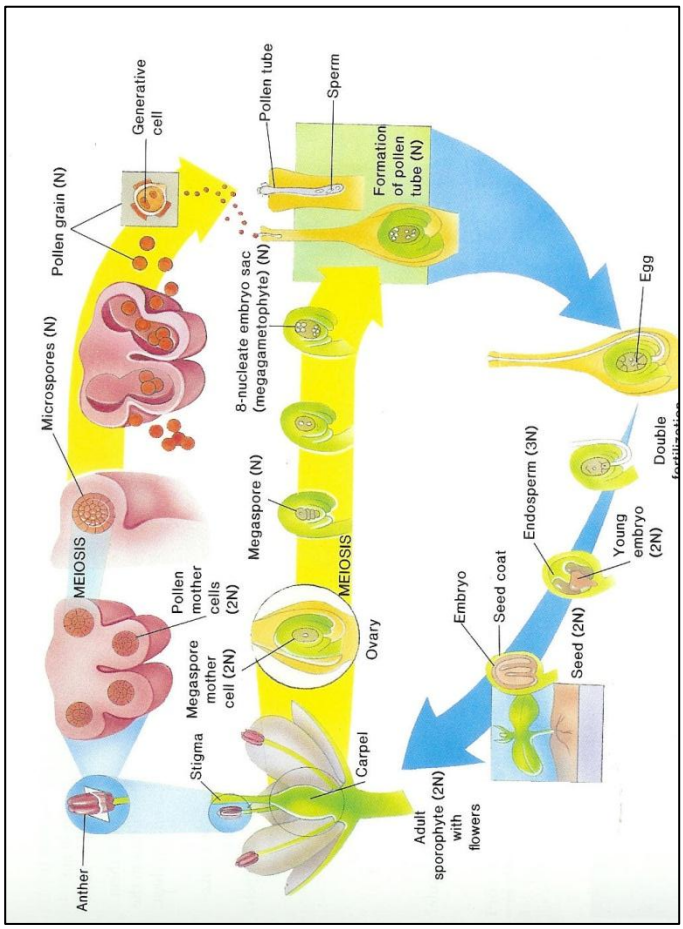
- a chromosomal condition that affects development in females. The most common feature of Turner syndrome is short stature, which becomes evident by about age 5. An early loss of ovarian function (ovarian

hypofunction or premature ovarian failure) is also very common. The ovaries develop normally at first, but egg cells (oocytes) usually die prematurely and most ovarian tissue degenerates before birth. Many affected girls do not undergo puberty unless they receive hormone therapy, and most are unable to conceive (infertile). A small percentage of females with Turner syndrome retain normal ovarian function through young adulthood.

- About 30 percent of females with Turner syndrome have extra folds of skin on the neck (webbedneck), a low hairline at the back of the neck, puffiness or swelling (lymphedema) of the hands and feet, skeletal abnormalities, or kidney problems. One third to one half of individuals with Turner syndrome are born with a heart defect, such as a narrowing of the large artery leaving the heart (coarctation of the aorta) or abnormalities of the valve that connects the aorta with the heart (the aortic valve). Complications associated with these heart defects can be life-threatening.
- Most girls and women with Turner syndrome have normal intelligence. Developmental delays, nonverbal learning disabilities, and behavioral problems are possible, although these characteristics vary among affected individuals

- This cond ition occurs in about 1 in 2,500 newborn girls worldwide, but it is much more common among pregnancies that do not survive to term (miscarriages and stillbirths).





Reference: https://ghr.nlm.nih.gov

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